

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	DiGeorge Syndrome	22q11.2 region	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	AAAS-related achalasia, alacrima, and adrenal insufficiency spectrum disorder	AAAS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Generalized arterial calcification of infancy	ABCC6-related pseudoxanthoma elasticum spectrum disorder	ABCC6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	ABCC8-related hyperinsulinemic hypoglycemia	ABCC8	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	ABCC8-related neonatal diabetes	ABCC8	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Disorder of peroxisomal fatty acid oxidation	ABCD1-related adrenoleukodystrophy	ABCD1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	ABCD4-related methylmalonic aciduria and homocystinuria, cblJ type	ABCD4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of complex I subunits and assembly factors	ACAD9-related acyl-CoA dehydrogenase 9 deficiency	ACAD9	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial fatty acid oxidation	ACADM-related medium-chain acyl-CoA dehydrogenase deficiency	ACADM	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial fatty acid oxidation	ACADVL-related very long-chain acyl-CoA dehydrogenase deficiency	ACADVL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of ketone body metabolism	ACAT1-related alpha-methylacetoacetic aciduria	ACAT1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	ACD-related telomere biology disorder	ACD	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	ACP5-related skeletal dysplasia with immune dysregulation	ACP5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Smooth muscle dysfunction	ACTA2-related thoracic aortic aneurysm and smooth muscle dysfunction spectrum	ACTA2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Functional bowel disorders	ACTG2-related visceral myopathy	ACTG2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet

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Inborn errors of immunity	T-B- severe combined immune deficiency	ADA-related adenosine deaminase deficiency	ADA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	ADA2-related ADA2 deficiency	ADA2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	ADAM17-related inflammatory skin and bowel disease	ADAM17	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	ADAMTS13-related thrombotic thrombocytopenic purpura	ADAMTS13	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	ADGRV1-related Usher syndrome	ADGRV1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of the metabolism of sulfur-containing amino acids and hydrogen sulfide	ADK-related adenosine kinase deficiency	ADK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycogen metabolism	AGL-related glycogen storage disease 3	AGL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Lipodistrophy disorders	AGPAT2-related lipodystrophy	AGPAT2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	AGRN-related myasthenia	AGRN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Hyperoxaluria	AGXT-related primary hyperoxaluria	AGXT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of the metabolism of sulfur-containing amino acids and hydrogen sulfide	AHCY-related S-adenosylhomocysteine hydrolase deficiency	AHCY	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in serum IgG and IgA with normal/elevated IgM and normal numbers of B cells, hyper IgM	AICDA-related immunodeficiency	AICDA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation	AIRE-related autoimmune polyendocrinopathy disorder	AIRE	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B- severe combined immune deficiency	AK2-related reticular dysgenesis	AK2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Bile acid synthesis defects	AKR1D1-related bile acid synthesis defect	AKR1D1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

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Hematologic disorders	Erythropoietic porphyrias	ALAS2-related protoporphyria	ALAS2	XL	New function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of ornithine, proline and hydroxyproline metabolism	ALDH4A1-related hyperprolinemia	ALDH4A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyridoxine metabolism	ALDH7A1-related pyridoxine-dependent epilepsy	ALDH7A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of galactose and fructose metabolism	ALDOB-related hereditary fructose intolerance	ALDOB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	ALG14-congenital disorder of glycosylation	ALG14	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	ALG2-related congenital disorder of glycosylation spectrum	ALG2	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	ALK-related neuroblastic tumor susceptibility	ALK	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Hereditary anemias	AMN-related megaloblastic anemia	AMN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycine and serine metabolism	AMT-related glycine encephalopathy	AMT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	ANKRD17-related neurodevelopmental disorder with multiple anomalies	ANKRD17	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	ANOS1-related hypogonadotropic hypogonadism	ANOS1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of copper metabolism	AP1S1-related ichthyosis and multiple anomalies spectrum disorder	AP1S1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis syndromes with hypopigmentation	AP3B1-related Hermansky-Pudlak syndrome	AP3B1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis syndromes with hypopigmentation	AP3D1-related Hermansky-Pudlak syndrome	AP3D1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	APC-related familial adenomatous polyposis	APC	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet

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Gastrointestinal disorders	Disorders of lipoprotein metabolism	APOB-related hypobetalipoproteinemia	APOB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of purine metabolism	APRT-related adenine phosphoribosyltransferase deficiency	APRT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Diabetes insipidus	AQP2-related nephrogenic diabetes insipidus	AQP2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	ARG1-related arginase deficiency	ARG1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immunodeficiency with congenital thrombocytopenia	ARPC1B-related immune dysregulation disorder	ARPC1B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of sphingolipid degradation	ARSA-related arylsulfatase A deficiency	ARSA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycosaminoglycan degradation	ARSB-related mucopolysaccharidosis type 6	ARSB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	ASL-related argininosuccinic aciduria	ASL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	ASS1-related argininosuccinate synthetase deficiency	ASS1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	ATM-related ataxia-telangiectasia	ATM	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in at least 2 serum Ig isotypes with normal or low B cell numbers, CVID phenotype	ATP6AP1-related immunodeficiency	ATP6AP1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	ATP6AP2-related neurodevelopmental and movement disorder	ATP6AP2	XL	Unknown	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Renal tubular acidosis	ATP6V0A4-related distal renal tubular acidosis	ATP6V0A4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Renal tubular acidosis	ATP6V1B1-related renal tubular acidosis with hearing loss	ATP6V1B1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of copper metabolism	ATP7A-related Menkes spectrum disorder	ATP7A	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet

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Endocrine disorders	Gonadal dysgenesis disorders	ATRX-related alpha-thalassemia and intellectual disability spectrum disorder	ATRX	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Diabetes insipidus	AVP-related diabetes insipidus	AVP	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Diabetes insipidus	AVPR2-related nephrogenic diabetes insipidus	AVPR2	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Bile acid synthesis defects	BAAT-related bile acid defect	BAAT	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of branched-chain amino acid metabolism	BCKDHA-related maple syrup urine disease	BCKDHA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of branched-chain amino acid metabolism	BCKDHB-related maple syrup urine disease	BCKDHB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of branched-chain amino acid metabolism	BCKDK-related branched-chain keto acid dehydrogenase kinase deficiency	BCKDK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	BCL10-related immunodeficiency	BCL10	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	BCL11B-related neurodevelopmental disorder	BCL11B	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	BLNK-related agammaglobulinemia	BLNK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	BMP1-related osteogenesis imperfecta	BMP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	BMP4-related multiple anomalies spectrum disorder	BMP4	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	BRAF-related RASopathy	BRAF	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Lipodistrophy disorders	BSCL2-related lipodystrophy	BSCL2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Salt-wasting tubulopathies	BSND-related Bartter syndrome	BSND	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

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Inherited metabolic disorders	Disorder of biotin metabolism	BTD-related biotinidase deficiency	BTD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	BTK-related agammaglobulinemia	BTK	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C1QA-related complement factor deficiency	C1QA	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C1QB-related complement factor deficiency	C1QB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C1QC-related C1q deficiency	C1QC	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C2-related complement factor deficiency	C2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C3-related complement factor deficiency	C3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C5-related complement factor deficiency	C5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C6-related complement factor deficiency	C6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C7-related complement factor deficiency	C7	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C8A-related complement factor deficiency	C8A	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C8B-related complement factor deficiency	C8B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	C9-related complement factor deficiency	C9	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	CA5A-related carbonic anhydrase VA deficiency	CA5A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	CACNA1C-related arrhythmogenic disorder	CACNA1C	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet

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Endocrine disorders	Mineralocorticoid disorders	CACNA1D-related primary aldosteronism and neurodevelopmental spectrum disorder	CACNA1D	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	CAD-related epilepsy and neurodevelopmental disorder	CAD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Isotype, light Chain, or functional deficiencies with generally normal numbers of B cells	CARD11-related immune dysregulation disorder	CARD11	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	CARD11-related immunodeficiency	CARD11	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	CARD11-related polyclonal B-cell lymphocytosis	CARD11	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to invasive fungal diseases	CARD9-related candidiasis	CARD9	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Susceptibility to EBV and lymphoproliferative conditions	CARMIL2-related immune dysregulation disorder	CARMIL2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	CASQ2-related catecholaminergic polymorphic ventricular tachycardia	CASQ2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Parathyroid disorders	CASR-related hyperparathyroidism	CASR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Lipodistrophy disorders	CAV1-related lipodystrophy	CAV1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Lipodistrophy disorders	CAVIN1-related lipodystrophy	CAVIN1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation	CBLB-related immune dysregulation disorder	CBLB	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	CBLIF-related megaloblastic anemia	CBLIF	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of the metabolism of sulfur-containing amino acids and hydrogen sulfide	CBS-related homocystinuria	CBS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	CCBE1-related Hennekam lymphangiectasia-lymphedema syndrome	CCBE1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet

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Inborn errors of immunity	Susceptibility to EBV and lymphoproliferative conditions	CD27-related immunodeficiency	CD27	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	CD3D-related immunodeficiency	CD3D	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	CD3E-related immunodeficiency	CD3E	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	CD3G-related immunodeficiency	CD3G	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	CD40-related immunodeficiency	CD40	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	CD40LG-related immunodeficiency	CD40LG	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	CD55-related complement hyperactivation with protein-losing enteropathy	CD55	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	CD59-related CD59 deficiency	CD59	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Susceptibility to EBV and lymphoproliferative conditions	CD70-related lymphoproliferative disorder	CD70	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	CD79A-related agammaglobulinemia	CD79A	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	CD79B-related agammaglobulinemia	CD79B	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	CDC42-related neurodevelopmental disorder with multiple anomalies	CDC42	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet

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Inborn errors of immunity	Other DNA repair defects	CDCA7-related immunodeficiency and multiple anomalies spectrum disorder	CDCA7	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	CDH23-related Usher syndrome	CDH23	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	CDON-related ocular defects	CDON	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet

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Inborn errors of immunity	Congenital neutropenias	CEBPE-related immune dysregulation disorder	CEBPE	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	CFD-related complement factor deficiency	CFD	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Complement deficiency syndromes	CFP-related properdin deficiency	CFP	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Pulmonary disorders	Cystic fibrosis	CFTR-related cystic fibrosis and congenital absence of the vas deferens	CFTR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	CHAT-related myasthenia	CHAT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Thymic defects with additional congenital anomalies	CHD7-related CHARGE spectrum disorder	CHD7	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	CHRNA1-related myasthenia	CHRNA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	CHRNA1-related myasthenia	CHRNA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	CHRNA1-related myasthenia	CHRNA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	CHRNA1-related myasthenia	CHRNA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	CHRNA1-related myasthenia	CHRNA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	CHRNA1-related myasthenia	CHRNA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	CHUK-related multiple congenital anomalies and immunodeficiency spectrum disorder	CHUK	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	CIB2-related hearing loss	CIB2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	CIITA-related major histocompatibility complex class II deficiency	CIITA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Mineralocorticoid disorders	CLCN2-related hyperaldosteronism	CLCN2	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	CLCN7-related osteopetrosis	CLCN7	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

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Renal disorders	Salt-wasting tubulopathies	CLCNKB-related Bartter syndrome	CLCNKB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	CLPB-related caseinolytic peptidase B deficiency	CLPB	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	CLPB-related caseinolytic peptidase B deficiency	CLPB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	CLRN1-related Usher syndrome	CLRN1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	COL13A1-related myasthenia	COL13A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	COL1A1-related arthrochalasia Ehlers-Danlos syndrome	COL1A1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	COL1A1-related osteogenesis imperfecta and Caffey disease	COL1A1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	COL1A1-related osteogenesis imperfecta	COL1A1	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	COL1A1-related osteogenesis imperfecta-Ehlers-Danlos spectrum disorder	COL1A1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	COL1A2-related arthrochalasia Ehlers-Danlos syndrome	COL1A2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	COL1A2-related osteogenesis imperfecta	COL1A2	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	COL1A2-related osteogenesis imperfecta-Ehlers-Danlos spectrum disorder	COL1A2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	COLQ-related myasthenia	COLQ	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	COPA-related autoimmune interstitial lung, joint, and kidney disease	COPA	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of coenzyme Q10 biosynthesis	COQ2-related coenzyme Q10 deficiency	COQ2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

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Inherited metabolic disorders	Disorders of coenzyme Q10 biosynthesis	COQ4-related coenzyme Q10 deficiency	COQ4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of coenzyme Q10 biosynthesis	COQ6-related coenzyme Q10 deficiency	COQ6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of coenzyme Q10 biosynthesis	COQ7-related coenzyme Q10 deficiency	COQ7	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of coenzyme Q10 biosynthesis	COQ8A-related coenzyme Q10 deficiency	COQ8A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Nephrotic syndrome	COQ8B-related nephrotic syndrome	COQ8B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of coenzyme Q10 biosynthesis	COQ9-related coenzyme Q10 deficiency	COQ9	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	CORO1A-related immunodeficiency	CORO1A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	CPS1-related carbamoylphosphate synthetase I deficiency	CPS1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of carnitine metabolism	CPT1A-related carnitine palmitoyltransferase 1A deficiency	CPT1A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of carnitine metabolism	CPT2-related carnitine palmitoyltransferase deficiency	CPT2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	CSF3R-related neutropenia	CSF3R	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	CTC1-related telomere biology disorder	CTC1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Regulatory T cell defects	CTLA4-related immune dysregulation	CTLA4	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Cystinosis	CTNS-related cystinosis	CTNS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Susceptibility to EBV and lymphoproliferative conditions	CTPS1-related immunodeficiency	CTPS1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	CTR9-related Wilms tumor	CTR9	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	CTSC-related syndromic and nonsyndromic periodontitis	CTSC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Hereditary anemias	CUBN-related megaloblastic anemia	CUBN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	CUL3-related pseudohypoaldosteronism	CUL3	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Epidermodysplasia verruciformis (HPV)	CXCR4-related immunodeficiency and heart defects spectrum disorder	CXCR4	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of respiratory burst	CYBA-related chronic granulomatous disease	CYBA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of respiratory burst; Mendelian susceptibility to mycobacterial disease (MSMD)	CYBB-related chronic granulomatous disease	CYBB	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of respiratory burst	CYBC1-related chronic granulomatous disease	CYBC1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	CYP11A1-related congenital adrenal insufficiency	CYP11A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	CYP17A1-related 17-alpha-hydroxylase/17,20-lyase deficiency	CYP17A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Parathyroid disorders	CYP24A1-related hypercalcemia	CYP24A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Lipid and sterol metabolism disorders	CYP27A1-related cerebrotendinous xanthomatosis	CYP27A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Rickets - vitamin D and phosphate disorders	CYP2R1-related vitamin D 25-hydroxylase deficiency	CYP2R1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Bile acid synthesis defects	CYP7B1-related oxysterol 7 α -hydroxylase deficiency	CYP7B1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of branched-chain amino acid metabolism	DBT-related maple syrup urine disease	DBT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B- severe combined immune deficiency	DCLRE1C-related immunodeficiency	DCLRE1C	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Monoamine neurotransmission	DDC-related aromatic L-amino acid decarboxylase deficiency	DDC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Regulatory T cell defects	DEF6-related immune dysregulation disorder	DEF6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	DGAT1-related protein-losing enteropathy	DGAT1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	DGKE-related nephropathy	DGKE	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of folate metabolism	DHFR-related dihydrofolate reductase deficiency	DHFR	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	DHH-related variations of sex characteristics	DHH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	DHX37-related male gonadal dysgenesis	DHX37	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	DIAPH1-related neurodevelopmental disorder	DIAPH1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	DICER1-related pleuropulmonary blastoma familial tumor predisposition syndrome	DICER1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	DKC1-related telomere biology disorder	DKC1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyruvate metabolism	DLAT-related pyruvate dehydrogenase deficiency	DLAT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyruvate metabolism	DLD-related dihydrolipoamide dehydrogenase deficiency	DLD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Muscular dystrophy	DMD-related muscular dystrophy	DMD	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	DMP1-related hypophosphatemic rickets	DMP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Monoamine neurotransmission	DNAJC12-related hyperphenylalaninemia	DNAJC12	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	DNAJC21-related bone marrow failure disorder	DNAJC21	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	DNASE1L3-related systemic lupus erythematosus	DNASE1L3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	DNMT3B-related immunodeficiency and multiple anomalies spectrum disorder	DNMT3B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immune dysregulation with colitis	DOCK11-related immune dysregulation disorder	DOCK11	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	DOCK2-related immunodeficiency	DOCK2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	DOCK8-related immunodeficiency syndrome	DOCK8	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	DOK7-related myasthenia	DOK7	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	DPAGT1-related congenital disorder of glycosylation (CDG Ij)	DPAGT1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	DUOXA2-related hypothyroidism	DUOXA2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	ECHS1-related mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	ECHS1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	EFL1-related Shwachman-Diamond syndrome	EFL1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	EIF2AK3-related diabetes	EIF2AK3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	EIF2S3-related neurodevelopmental disorder with multiple anomalies	EIF2S3	XL	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	ELANE-related neutropenia	ELANE	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immune dysregulation with colitis	ELF4-related autoinflammatory disorder	ELF4	XL	Unknown	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Generalized arterial calcification of infancy	ENPP1-related ectopic mineralization disorder	ENPP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	EPCAM-related congenital tufting enteropathy	EPCAM	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	EPG5-related neurodevelopmental disorder with multiple anomalies	EPG5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial fatty acid oxidation	ETFA-related multiple acyl-CoA dehydrogenase deficiency	ETFA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial fatty acid oxidation	ETFB-related multiple acyl-CoA dehydrogenase deficiency	ETFB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial fatty acid oxidation	ETFDH-related multiple acyl-CoA dehydrogenase deficiency	ETFDH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of the metabolism of sulfur-containing amino acids and hydrogen sulfide	ETHE1-related ethylmalonic encephalopathy	ETHE1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immuno-osseous dysplasias	EXTL3-related skeletal dysplasia with immunodeficiency	EXTL3	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F10-related factor X deficiency	F10	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F11-related factor XI deficiency	F11	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F12-related factor XII deficiency	F12	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F13A1-related factor XIII deficiency	F13A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F13B-related factor XIII deficiency	F13B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F2-related prothrombin deficiency	F2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F5-related deficiency	F5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F7-related factor VII deficiency	F7	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F8-related hemophilia	F8	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	F9-related hemophilia	F9	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmune lymphoproliferative syndrome (ALPS)	FADD-related immunodeficiency with multiple anomalies	FADD	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of phenylalanine and tyrosine metabolism	FAH-related tyrosinemia	FAH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Parathyroid disorders	FAM111A-related skeletal dysplasia	FAM111A	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmune lymphoproliferative syndrome (ALPS)	FAS-related autoimmune lymphoproliferative syndrome	FAS	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmune lymphoproliferative syndrome (ALPS)	FASLG-related autoimmune lymphoproliferative syndrome	FASLG	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of gluconeogenesis	FBP1-related fructose-1,6-bisphosphatase deficiency	FBP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	FCHO1-related immunodeficiency	FCHO1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Erythropoietic porphyrias	FECH-related erythropoietic protoporphyria	FECH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of motility	FERMT3-related leukocyte adhesion deficiency	FERMT3	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	FGA-related congenital fibrinogen deficiency	FGA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	FGB-related congenital fibrinogen deficiency	FGB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	FGF23-related hypophosphatemic rickets	FGF23	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	FGFR1-related hypogonadotropic hypogonadism	FGFR1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Skeletal dysplasia	FGFR3-related skeletal dysplasia	FGFR3	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	FGG-related fibrinogen deficiency	FGG	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	FKBP10-related osteogenesis imperfecta	FKBP10	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of riboflavin metabolism	FLAD1-related flavin adenine dinucleotide synthetase deficiency	FLAD1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	FNIP1-related immunodeficiency with cardiomyopathy spectrum disorder	FNIP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Folate metabolism disorder	FOLR1-related cerebral folate transport deficiency	FOLR1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	FOXA2-related multiple anomalies	FOXA2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	FOXE1-related hypothyroidism with craniofacial anomalies	FOXE1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Thymic defects with additional congenital anomalies	FOXN1-related immunodeficiency	FOXN1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Thymic defects with additional congenital anomalies	FOXN1-related lymphopenia	FOXN1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Regulatory T cell defects	FOXP3-related immunodysregulation, polyendocrinopathy, and enteropathy	FOXP3	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycoprotein degradation	FUCA1-related alpha-L-fucosidase deficiency	FUCA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of N-linked protein glycosylation	FUT8-related congenital disorder of glycosylation	FUT8	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycogen metabolism	G6PC1-related glycogen storage disease 1	G6PC1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	G6PC3-related neutropenia spectrum disorder	G6PC3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Other disorders of complex molecule degradation	GAA-related glycogen storage disease 2	GAA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of sphingolipid degradation	GALC-related Krabbe disease	GALC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of galactose and fructose metabolism	GALK1-related galactokinase deficiency	GALK1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of galactose and fructose metabolism	GALM-related galactosemia	GALM	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycosaminoglycan degradation	GALNS-related mucopolysaccharidosis type 4	GALNS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	GALNT3-related congenital disorder of glycosylation	GALNT3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of galactose and fructose metabolism	GALT-related galactosemia	GALT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of creatine metabolism	GAMT-related guanidinoacetate methyltransferase deficiency	GAMT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Syndromic disorders	GATA1-related cytopenia	GATA1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other non-lymphoid defects	GATA2-related GATA2 deficiency	GATA2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Parathyroid disorders	GATA3-related hypoparathyroidism, hearing loss, and renal dysplasia	GATA3	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	GATA6-related multiple anomalies	GATA6	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of creatine metabolism	GATM-related arginine:glycine amidinotransferase deficiency	GATM	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	GCDH-related glutaryl-CoA dehydrogenase deficiency	GCDH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of tetrahydrobiopterin metabolism	GCH1-related cyclohydrolase 1 deficiency	GCH1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	GCK-related hyperinsulinemic hypoglycemia	GCK	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	GCK-related neonatal diabetes	GCK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Parathyroid disorders	GCM2-related hyperparathyroidism or hypoparathyroidism	GCM2	AD	Loss of function and gain of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	GFPT1-related myasthenia	GFPT1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	GGCX-related vitamin K-dependent coagulation factor deficiency	GGCX	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Growth disorders	GHR-related growth hormone insensitivity	GHR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	GJB2-related sensorineural hearing loss	GJB2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycine and serine metabolism	GLDC-related glycine encephalopathy	GLDC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	GLI2-related holoprosencephaly spectrum disorder	GLI2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	GLI3-related Pallister-Hall syndrome	GLI3	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	GLIS3-related diabetes mellitus and hypothyroidism spectrum disorder	GLIS3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Hyperekplexia	GLRA1-related hyperekplexia	GLRA1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Hyperekplexia	GLRA1-related hyperekplexia	GLRA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Hyperekplexia	GLRB-related hyperekplexia	GLRB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	GLUD1-related familial hyperinsulinism-hyperammonemia	GLUD1	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	GMPPB-related dystroglycanopathy	GMPPB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Parathyroid disorders	GNA11-related hypocalcemia	GNA11	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial shuttles and carriers	GOT2-related mitochondrial glutamate oxaloacetate transaminase deficiency	GOT2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	GP1BA-related Bernard-Soulier syndrome	GP1BA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	GP1BB-related Bernard-Soulier syndrome	GP1BB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	GP9-related Bernard-Soulier syndrome	GP9	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	GPC3-related Simpson-Golabi-Behmel syndrome	GPC3	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Disorders of lipoprotein metabolism	GPIHBP1-related hypercholesterolemia	GPIHBP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Hyperoxaluria	GRHPR-related primary hyperoxaluria	GRHPR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	GUCY2C-related gastrointestinal disorder	GUCY2C	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycosaminoglycan degradation	GUSB-related mucopolysaccharidosis type 7	GUSB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycogen metabolism	GYS2-related glycogen storage disease	GYS2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial fatty acid oxidation	HADH-related 3-hydroxyacyl-CoA dehydrogenase deficiency	HADH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

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Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial fatty acid oxidation	HADHA-related long chain hydroxyacyl-CoA dehydrogenase deficiency	HADHA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial fatty acid oxidation	HADHB-related mitochondrial trifunctional protein deficiency	HADHB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	HAX1-related neutropenia	HAX1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Hemoglobinopathy	HBB-related sickle cell disease	HBB	AR	New function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	HESX1-related pituitary hormone deficiency	HESX1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	HIBCH-related 3-hydroxyisobutryl-CoA hydrolase deficiency	HIBCH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of biotin metabolism	HLCS-related holocarboxylase synthetase deficiency	HLCS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of ketone body metabolism	HMGCL-related HMG-CoA lyase deficiency	HMGCL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Hyperoxaluria	HOGA1-related primary hyperoxaluria	HOGA1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of phenylalanine and tyrosine metabolism	HPD-related tyrosinemia	HPD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	HRAS-related RASopathy	HRAS	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Mineralocorticoid disorders	HSD11B2-related apparent mineralocorticoid excess	HSD11B2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	HSD3B2-related 3-beta-hydroxysteroid dehydrogenase 2 deficiency	HSD3B2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Bile acid synthesis defects	HSD3B7-related bile acid synthesis defect	HSD3B7	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycosaminoglycan degradation	IDS-related mucopolysaccharidosis type 2	IDS	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

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Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycosaminoglycan degradation	IDUA-related mucopolysaccharidosis type 1	IDUA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	IER3IP1-related brain malformations and diabetes mellitus	IER3IP1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	IFITM5-related osteogenesis imperfecta	IFITM5	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to severe viral infection	IFNAR1-related immunodeficiency	IFNAR1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to severe viral infection	IFNAR2-related immunodeficiency	IFNAR2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Mendelian susceptibility to mycobacterial disease (MSMD)	IFNGR1-related immunodeficiency	IFNGR1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Mendelian susceptibility to mycobacterial disease (MSMD)	IFNGR2-related immunodeficiency	IFNGR2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Growth disorders	IGF1-related insulin-like growth factor I deficiency	IGF1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	IGLL1-related immunodeficiency	IGLL1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	IGSF1-related central hypothyroidism and macroorchidism	IGSF1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Anhidrotic ectodermodyplasia with immunodeficiency (EDA-ID)	IKBKB-related immunodeficiency	IKBKB	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	IKBKB-related immunodeficiency	IKBKB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in at least 2 Serum Ig isotypes with normal or low B cell numbers, CVID phenotype	IKZF1-related immune dysregulation disorder	IKZF1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immune dysregulation with colitis	IL10RA-related inflammatory bowel disease	IL10RA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immune dysregulation with colitis	IL10RB-related inflammatory bowel disease	IL10RB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Mendelian susceptibility to mycobacterial disease (MSMD)	IL12B-related immunodeficiency	IL12B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Mendelian susceptibility to mycobacterial disease (MSMD)	IL12RB1-related immunodeficiency	IL12RB1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to mucocutaneous candidiasis	IL17RA-related candidiasis	IL17RA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to mucocutaneous candidiasis	IL17RC-related candidiasis	IL17RC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	IL1RN-related interleukin 1 receptor antagonist deficiency	IL1RN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Regulatory T cell defects	IL2RA-related immune dysregulation disorder	IL2RA	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Regulatory T cell defects	IL2RB-related immune dysregulation disorder	IL2RB	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	IL2RG-related immune dysregulation disorder	IL2RG	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	IL36RN-related pustular psoriasis	IL36RN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	IL6R-related immunodeficiency	IL6R	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	IL6ST-related immunodeficiency with skeletal anomalies	IL6ST	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	IL6ST-related immunodeficiency with skeletal anomalies	IL6ST	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	IL7R-related immunodeficiency	IL7R	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	INS-related neonatal diabetes	INS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	TLR signaling pathway deficiency	IRAK4-related immunodeficiency	IRAK4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	IRF4-related immunodeficiency	IRF4	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Mendelian susceptibility to mycobacterial disease (MSMD)	IRF8-related immunodeficiency	IRF8	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	IRS4-related hypothyroidism	IRS4	XL	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Mendelian susceptibility to mycobacterial disease (MSMD)	ISG15-related immunodeficiency	ISG15	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation	ITCH-related neurodevelopmental and autoimmune disorder	ITCH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	ITGA2B-related Glanzmann thrombasthenia	ITGA2B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of motility	ITGB2-related leukocyte adhesion deficiency	ITGB2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Bleeding disorders	ITGB3-related Glanzmann thrombasthenia	ITGB3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	ITK-related lymphoproliferative syndrome	ITK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Calcium channel defects	ITPR3-related immunodeficiency syndrome	ITPR3	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	IVD-related isovaleric acidemia	IVD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	IYD-related hypothyroidism	IYD	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Cholestasis disorders	JAG1-related Alagille syndrome	JAG1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	JAGN1-related neutropenia	JAGN1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation; Phenocopies of inborn errors of immunity	JAK1-related immune dysregulation disorder	JAK1	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	JAK3-related severe combined immune deficiency	JAK3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	KCNH2-related long QT syndrome	KCNH2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Salt-wasting tubulopathies	KCNJ1-related Bartter syndrome	KCNJ1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	KCNJ11-related hyperinsulinemic hypoglycemia	KCNJ11	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Mineralocorticoid disorders	KCNJ5-related hyperaldosteronism	KCNJ5	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	KCNQ1-related Jervell and Lange-Nielsen syndrome	KCNQ1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	KCNQ1-related LQTS arrhythmogenic spectrum disorder	KCNQ1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	KCNQ2-related epilepsy	KCNQ2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	KCNQ3-related epilepsy	KCNQ3	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	KDELR2-related osteogenesis imperfecta	KDELR2	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	KDM6A-related Kabuki syndrome	KDM6A	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	KLHL3-related pseudohypoaldosteronism	KLHL3	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	KLHL3-related pseudohypoaldosteronism	KLHL3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	KMT2D-related Kabuki syndrome	KMT2D	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	KRAS-related RASopathy	KRAS	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation	LACC1-related juvenile arthritis	LACC1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	LAT-related immunodeficiency	LAT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	LCK-related immunodeficiency	LCK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	LCP2-related immunodeficiency	LCP2	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	LCT-related lactase deficiency	LCT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Obesity disorders	LEP-related leptin deficiency	LEP	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Obesity disorders	LEPR-related obesity	LEPR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	LHX3-related combined pituitary hormone deficiency	LHX3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	LHX4-related combined pituitary hormone deficiency	LHX4	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	LIG1-related immunodeficiency	LIG1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B- severe combined immune deficiency	LIG4-related DNA ligase IV deficiency	LIG4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Other disorders of complex molecule degradation	LIPA-related cholesteryl ester storage disease	LIPA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	LMBRD1-related methylmalonic aciduria and homocystinuria, cblF type	LMBRD1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	LPIN2-related multi-system inflammatory disorder	LPIN2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Lipid disorders	LPL-related lipoprotein lipase deficiency	LPL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Regulatory T cell defects	LRBA-related immune dysregulation disorder	LRBA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	LRP5-related ocular and skeletal anomalies	LRP5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis syndromes with hypopigmentation	LYST-related Chediak-Higashi syndrome	LYST	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	LZTR1-related RASopathy	LZTR1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	MADD-related neurodevelopmental disorder	MADD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Salt-wasting tubulopathies	MAGED2-related Bartter syndrome	MAGED2	XL	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	MALT1-related immunodeficiency	MALT1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	MAMLD1-related variations of sex characteristics	MAMLD1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycoprotein degradation	MAN2B1-related alpha-mannosidosis	MAN2B1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	MAP2K1-related RASopathy	MAP2K1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	MAP3K1-related variations of sex characteristics	MAP3K1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	MC2R-related glucocorticoid deficiency	MC2R	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	MCCC1-related 3-methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	MCCC2-related 3-methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	MCEE-related methylmalonyl-CoA epimerase deficiency	MCEE	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	MCM4-related immunodeficiency	MCM4	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	MECOM-related bone marrow failure and skeletal anomalies spectrum disorder	MECOM	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Dysregulation of pyrin inflammasome	MEFV-related familial mediterranean fever	MEFV	AR	New function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	MESD-related osteogenesis imperfecta	MESD	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	MLH1-related constitutional mismatch repair deficiency	MLH1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	MLYCD-related malonyl-CoA decarboxylase deficiency	MLYCD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	MMAA-related methylmalonic aciduria	MMAA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	MMAB-related methylmalonic aciduria, cblB type	MMAB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	MMACHC-related methylmalonic aciduria and homocystinuria, cblC type	MMACHC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	MMADHC-related homocystinuria, cblD type	MMADHC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	MMUT-related methylmalonic aciduria	MMUT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of molybdenum cofactor metabolism	MOCS1-related molybdenum cofactor deficiency	MOCS1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in at least 2 serum Ig isotypes with normal or low B cell numbers, CVID phenotype	MOGS-related congenital disorder of glycosylation (CDG IIb)	MOGS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	Turner Syndrome	Monosomy X		Aneuploidy	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of N-linked protein glycosylation	MPI-related congenital disorder of glycosylation (CDG Ib)	MPI	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Hematologic bone marrow failure syndromes	MPL-related thrombocytopenia	MPL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	MRAP-related glucocorticoid deficiency	MRAP	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	MRAS-related RASopathy	MRAS	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	MSH2-related constitutional mismatch repair deficiency	MSH2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	MSH6-related constitutional mismatch repair deficiency	MSH6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	MSN-related immunodeficiency	MSN	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of vitamin B12 and folate metabolism	MTHFD1-related immunodeficiency and megaloblastic anemia	MTHFD1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of folate metabolism	MTHFR-related MTHFR deficiency	MTHFR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of the metabolism of sulfur-containing amino acids and hydrogen sulfide	MTR-related methylcobalamin deficiency, cblG type	MTR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	MTRR-related methionine synthase reductase deficiency, cbl E type	MTRR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Disorders of lipoprotein metabolism	MTTP-related abetalipoproteinemia	MTTP	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	MUSK-related myasthenia	MUSK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects affecting the inflammasome	MVK-related mevalonate kinase deficiency	MVK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	TLR signaling pathway deficiency	MYD88-related immunodeficiency	MYD88	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	MYO5B-related enteropathy and cholestasis spectrum disorder	MYO5B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	MYO7A-related Usher spectrum disorder	MYO7A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	MYRF-related cardiac and urogenital anomalies spectrum disorder	MYRF	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immuno-osseous dysplasias	MYSM1-related bone marrow failure disorder	MYSM1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	NAGS-related N-acetylglutamate synthase deficiency	NAGS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of multiple glycosylation pathways	NANS-related congenital disorder of glycosylation	NANS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	NBAS-related liver failure and multiple anomalies spectrum disorder	NBAS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Regulatory T cell defects	NBEAL2-related gray platelet syndrome	NBEAL2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	NBN-related Nijmegen breakage syndrome	NBN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of respiratory burst	NCF2-related chronic granulomatous disease	NCF2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of respiratory burst	NCF4-related chronic granulomatous disease	NCF4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	NCKAP1L-related immune dysregulation disorder	NCKAP1L	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	NEUROG3-related enteric anendocrinosis	NEUROG3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Multisystem disease	NF1-related neurofibromatosis	NF1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Anhidrotic ectodermodyplasia with immunodeficiency (EDA-ID)	NFKBIA-related ectodermal dysplasia and immunodeficiency spectrum disorder	NFKBIA	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B- severe combined immune deficiency	NHEJ1-related immunodeficiency and radiation sensitivity	NHEJ1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	NKX2-1-related movement disorder	NKX2-1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects affecting the inflammasome	NLRC4-related autoinflammatory disorder	NLRC4	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects affecting the inflammasome	NLRP12-related familial cold autoinflammatory syndrome	NLRP12	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects affecting the inflammasome	NLRP3-related autoinflammatory disorder	NLRP3	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	NNT-related glucocorticoid deficiency	NNT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	NOD2-related Blau syndrome	NOD2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Cholestasis disorders	NOTCH2-related Alagille syndrome	NOTCH2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Other disorders of complex molecule degradation	NPC1-related Niemann-Pick disease	NPC1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Other disorders of complex molecule degradation	NPC2-related Niemann-Pick disease	NPC2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Nephrotic syndrome	NPHS1-related nephrotic syndrome	NPHS1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	NR0B1-related congenital adrenal hypoplasia	NR0B1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Mineralocorticoid disorders	NR3C2-related pseudohypoaldosteronism	NR3C2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	NR5A1-related variations of sex characteristics	NR5A1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	NRAS-related RASopathy	NRAS	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

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Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	NSMCE3-related chromosome breakage disorder with lung disease	NSMCE3	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	OAS1-related immune dysregulation disorder	OAS1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of ornithine, proline and hydroxyproline metabolism	OAT-related gyrate atrophy of the choroid and retina	OAT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of polyamine metabolism	ODC1-related neurodevelopmental disorder with multiple anomalies	ODC1	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	OFD1-related ciliopathy	OFD1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Calcium channel defects	ORAI1-related immunodeficiency	ORAI1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	OSTM1-related osteopetrosis	OSTM1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	OTC-related ornithine transcarbamylase deficiency	OTC	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	OTOF-related nonsyndromic auditory neuropathy spectrum disorder	OTOF	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	OTULIN-related autoinflammatory disorder	OTULIN	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	OTX2-related ocular malformations and pituitary dysfunction	OTX2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of ketone body metabolism	OXCT1-related succinyl CoA:3-oxoacid CoA transferase deficiency	OXCT1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of phenylalanine and tyrosine metabolism	PAH-related phenylketonuria	PAH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	PARN-related telomere biology disorder	PARN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	PAX1-related otofaciocervical syndrome	PAX1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	PAX8-related hypothyroidism	PAX8	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of gluconeogenesis	PC-related pyruvate carboxylase deficiency	PC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of tetrahydrobiopterin metabolism	PCBD1-related BH4-deficient hyperphenylalaninemia	PCBD1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	PCCA-related propionic acidemia	PCCA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Organic acidurias	PCCB-related propionic acidemia	PCCB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	PCDH15-related Usher syndrome	PCDH15	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of gluconeogenesis	PCK1-related cytosolic phosphoenolpyruvate carboxykinase deficiency	PCK1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	PCSK1-related proprotein convertase 1 deficiency	PCSK1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyruvate metabolism	PDHA1-related pyruvate dehydrogenase deficiency	PDHA1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyruvate metabolism	PDHB-related pyruvate dehydrogenase deficiency	PDHB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyruvate metabolism	PDHX-related pyruvate dehydrogenase deficiency	PDHX	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyruvate metabolism	PDP1-related pyruvate dehydrogenase phosphatase deficiency	PDP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of coenzyme Q10 biosynthesis	PDSS1-related coenzyme Q10 deficiency	PDSS1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of coenzyme Q10 biosynthesis	PDSS2-related coenzyme Q10 deficiency	PDSS2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	PDX1-related pancreatic dysgenesis and diabetes spectrum disorder	PDX1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation	PEPD-related prolidase deficiency	PEPD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of multiple glycosylation pathways	PGM1-related congenital disorder of glycosylation (CDG It)	PGM1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	PGM3-related immunodeficiency	PGM3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Rickets - vitamin D and phosphate disorders	PHEX-related hypophosphatemic rickets	PHEX	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycine and serine metabolism	PHGDH-related phosphoglycerate dehydrogenase deficiency	PHGDH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycogen metabolism	PHKA2-related glycogen storage disease 9	PHKA2	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycogen metabolism	PHKB-related glycogen storage disease 9	PHKB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycogen metabolism	PHKG2-related glycogen storage disease 9	PHKG2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in at least 2 serum Ig isotypes with normal or low B cell numbers, CVID phenotype	PI4KA-related brain malformations spectrum disorder	PI4KA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in at least 2 serum Ig isotypes with normal or low B cell numbers, CVID phenotype	PIK3CD-related immunodeficiency	PIK3CD	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	PIK3CD-related immunodeficiency	PIK3CD	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	PIK3R1-related agammaglobulinemia	PIK3R1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in at least 2 serum Ig isotypes with normal or low B cell numbers, CVID phenotype	PIK3R1-related immunodeficiency	PIK3R1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in at least 2 serum Ig isotypes with normal or low B cell numbers, CVID phenotype	PIK3R1-related SHORT syndrome	PIK3R1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Hemolytic disorder	PKLR-related pyruvate kinase deficiency	PKLR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects affecting multiple immune pathways	PLCG2-related immune dysregulation disorder	PLCG2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	PLEC-related epidermolysis bullosa-muscular dystrophy spectrum disorder	PLEC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Ophthalmologic disorders	Plasminogen deficiency	PLG-related plasminogen deficiency	PLG	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyridoxine metabolism	PLPBP-related vitamin B6-dependent epilepsy	PLPBP	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	PLS3-related osteoporosis	PLS3	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	PLVAP-related protein-losing enteropathy	PLVAP	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	PNP-related purine nucleoside phosphorylase deficiency	PNP	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	PNPLA6-related disorder	PNPLA6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyridoxine metabolism	PNPO-related pyridoxamine 5'-phosphate oxidase deficiency	PNPO	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	POLA1-related reticulate pigmentary disorder	POLA1	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	POLE-related skeletal dysplasia	POLE	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mtDNA replication and maintenance	POLG-related disorder	POLG	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Dermatologic disorders	Xeroderma pigmentosum	POLH-related xeroderma pigmentosum	POLH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	POMC-related proopiomelanocortin deficiency	POMC	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	POMP-related immune dysregulation disorder	POMP	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	POU1F1-related combined pituitary hormone deficiency	POU1F1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	PPA2-related sudden cardiac failure	PPA2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	PPP2R3C-related myoectodermal gonadal dysgenesis syndrome	PPP2R3C	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of cobalamin metabolism	PRDX1-related disorder	PRDX1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	PREPL-related PREPL deficiency	PREPL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis	PRF1-related hemophagocytic lymphohistiocytosis	PRF1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	PRIM1-related DNA replication disorder	PRIM1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	PRKAR1A-related Carney complex spectrum disorder	PRKAR1A	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Susceptibility to EBV and lymphoproliferative conditions	PRKCD-related immune dysregulation disorder	PRKCD	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B- severe combined immune deficiency	PRKDC-related immune dysregulation disorder	PRKDC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	PROK2-related hypogonadotropic hypogonadism	PROK2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	PROKR2-related hypogonadotropic hypogonadism	PROKR2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	PROP1-related combined pituitary hormone deficiency	PROP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	PRRT2-related movement and neurodevelopmental spectrum disorder	PRRT2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	PRRT2-related movement and neurodevelopmental spectrum disorder	PRRT2	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions/Type I interferonopathy	PSMB8-related immune dysregulation disorder	PSMB8	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	PSMC3-related neurodevelopmental disorder with multiple anomalies	PSMC3	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	PSMC5-related neurodevelopmental disorder	PSMC5	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	PSTPIP1-related autoinflammatory disorder	PSTPIP1	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	PTCH1-related nevoid basal cell carcinoma syndrome	PTCH1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	PTF1A-related pancreatic agenesis	PTF1A	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Parathyroid disorders	PTH-related hypoparathyroidism	PTH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	PTPN11-related Noonan syndrome with multiple lentigines	PTPN11	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	PTPN11-related RASopathy	PTPN11	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	PTPN2-related immunodeficiency	PTPN2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B+ severe combined immune deficiency	PTPRC-related immunodeficiency	PTPRC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of tetrahydrobiopterin metabolism	PTS-related BH4-deficient hyperphenylalaninemia	PTS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of glycogen metabolism	PYGL-related glycogen storage disease 6	PYGL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of tetrahydrobiopterin metabolism	QDPR-related BH4-deficient hyperphenylalaninemia	QDPR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis syndromes with hypopigmentation	RAB27A-related Griscelli syndrome	RAB27A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B- severe combined immune deficiency	RAC2-related immunodeficiency	RAC2	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of motility	RAC2-related neutrophil immunodeficiency syndrome	RAC2	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	RAF1-related RASopathy	RAF1	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B- severe combined immune deficiency	RAG1-related immune dysregulation	RAG1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	T-B- severe combined immune deficiency	RAG2-related immune dysregulation disorder	RAG2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	RANBP2-related acute necrotizing encephalopathy	RANBP2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	RAPSN-related myasthenia	RAPSN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Susceptibility to EBV and lymphoproliferative conditions	RASGRP1-related immunodeficiency	RASGRP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	RAX-related microphthalmia	RAX	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	RB1-related retinoblastoma	RB1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	RBCK1-related polyglucosan body myopathy	RBCK1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Hematologic bone marrow failure syndromes	RBM8A-related thrombocytopenia-absent radius syndrome	RBM8A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	RECQL4-related skeletal dysplasia	RECQL4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	NF-kB mediated/ Type 1 interferonopathies	RELA-related immune dysregulation disorder	RELA	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	RELA-related immune dysregulation disorder	RELA	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	RELB-related immunodeficiency	RELB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	RET-related multiple endocrine neoplasia 2B	RET	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	RFX5-related immunodeficiency	RFX5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	RFX6-related neonatal diabetes	RFX6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	RFXANK-related major histocompatibility complex class II deficiency	RFXANK	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	RFXAP-related major histocompatibility complex class II deficiency	RFXAP	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects affecting the inflammasome	RIPK1-related autoinflammatory disorder	RIPK1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immune dysregulation with colitis	RIPK1-related immunodeficiency	RIPK1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	RIT1-related RASopathy	RIT1	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immuno-osseous dysplasias	RMRP-related cartilage-hair hypoplasia - anauxetic dysplasia spectrum disorder	RMRP	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	RNF168-related immunodeficiency and radiation sensitivity	RNF168	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	RNF31-related immunodeficiency with autoinflammation	RNF31	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	RNPC3-related pituitary hypoplasia and growth deficiency	RNPC3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	ROBO1-related pituitary stalk interruption syndrome	ROBO1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Ophthalmologic disorders	Retinitis pigmentosa	RPE65-related retinal dystrophy	RPE65	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPL11-related Diamond-Blackfan anemia	RPL11	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPL15-related Diamond-Blackfan anemia	RPL15	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPL26-related Diamond-Blackfan anemia	RPL26	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPL35A-related Diamond-Blackfan anemia	RPL35A	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPL5-related Diamond-Blackfan anemia	RPL5	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPS10-related Diamond-Blackfan anemia	RPS10	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPS19-related Diamond-Blackfan anemia	RPS19	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPS24-related Diamond-Blackfan anemia	RPS24	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPS26-related Diamond-Blackfan anemia	RPS26	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPS29-related Diamond-Blackfan anemia	RPS29	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Diamond-Blackfan anemia	RPS7-related Diamond-Blackfan anemia	RPS7	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	RPSA-related congenital asplenia	RPSA	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	RRAS2-related RASopathy	RRAS2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	RTEL1-related telomere biology disorder	RTEL1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Pharmacologic disorders	RYR1-related malignant hyperthermia	RYR1	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	RYR2-related arrhythmogenic disorder	RYR2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	SAMD9-related MIRAGE syndrome	SAMD9	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	SAMD9L-related ataxia-pancytopenia syndrome	SAMD9L	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Disorders of lipoprotein metabolism	SAR1B-related chylomicron retention disease	SAR1B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	SCN1A-related developmental, epileptic encephalopathy and movement disorder	SCN1A	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	SCN5A-related arrhythmogenic disorder	SCN5A	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	SCN5A-related arrhythmogenic disorder	SCN5A	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	SCN8A-related epilepsy	SCN8A	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Mineralocorticoid disorders	SCNN1A-related pseudohypoaldosteronism	SCNN1A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Mineralocorticoid disorders	SCNN1B-related pseudohypoaldosteronism	SCNN1B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Mineralocorticoid disorders	SCNN1G-related pseudohypoaldosteronism	SCNN1G	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	SERPINF1-related osteogenesis imperfecta	SERPINF1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	SERPINH1-related osteogenesis imperfecta	SERPINH1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Pulmonary disorders	Surfactant deficiency	SFTPC-related surfactant metabolism dysfunction	SFTPC	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	SGPL1-related sphingosine phosphate lyase insufficiency syndrome	SGPL1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation	SH2B3-related myeloproliferative and autoimmune spectrum disorder	SH2B3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Susceptibility to EBV and lymphoproliferative conditions	SH2D1A-related lymphoproliferative disorder	SH2D1A	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	SHH-related holoprosencephaly spectrum disorder	SHH	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	SI-related sucrase-isomaltase deficiency	SI	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	SKIV2L-related trichohepatoenteric syndrome	SKIC2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	SKIC3-related trichohepatoenteric syndrome	SKIC3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Salt-wasting tubulopathies	SLC12A1-related Bartter syndrome	SLC12A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Salt-wasting tubulopathies	SLC12A3-related Gitelman syndrome	SLC12A3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	SLC13A5-related developmental and epileptic encephalopathy	SLC13A5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	SLC18A3-related myasthenia	SLC18A3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	SLC19A2-related thiamine-responsive megaloblastic anemia	SLC19A2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of thiamine metabolism	SLC19A3-related thiamine metabolism dysfunction	SLC19A3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of carnitine metabolism	SLC22A5-related primary systemic carnitine deficiency	SLC22A5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	SLC25A1-related D-2- and L-2-hydroxyglutaric aciduria	SLC25A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	SLC25A13-related citrullinemia	SLC25A13	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Urea cycle disorders and inherited hyperammonemias	SLC25A15-related hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	SLC25A15	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of carnitine metabolism	SLC25A20-related carnitine-acylcarnitine translocase deficiency	SLC25A20	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Hereditary anemias	SLC25A38-related sideroblastic anemia	SLC25A38	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	SLC26A3-related secretory diarrhea	SLC26A3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	SLC26A4-related hearing loss	SLC26A4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	SLC29A3-related histiocytosis-lymphadenopathy plus syndrome	SLC29A3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	SLC2A1-related GLUT1 deficiency	SLC2A1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	SLC2A2-related Fanconi-Bickel syndrome	SLC2A2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	SLC34A1-related infantile hypercalcemia	SLC34A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	SLC34A3-related hypophosphatemic rickets	SLC34A3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of multiple glycosylation pathways	SLC35A2-related congenital disorder of glycosylation (CDG II _m)	SLC35A2	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of motility	SLC35C1-related congenital disorder of glycosylation (CDG II _c)	SLC35C1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	SLC37A4-related glycogen storage disease 1	SLC37A4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of manganese metabolism	SLC39A14-related hypermanganesemia	SLC39A14	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of zinc metabolism	SLC39A4-related acrodermatitis enteropathica zinc-deficient	SLC39A4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	SLC39A7-related immunodeficiency	SLC39A7	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of manganese metabolism	SLC39A8-related congenital disorder of glycosylation (CDG II _n)	SLC39A8	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of vitamin B12 and folate metabolism	SLC46A1-related hereditary folate malabsorption	SLC46A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Renal tubular acidosis	SLC4A1-related renal tubular acidosis	SLC4A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Riboflavin transport disorders	SLC52A2-related riboflavin transporter deficiency	SLC52A2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Riboflavin transport disorders	SLC52A3-related riboflavin transporter deficiency	SLC52A3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	SLC5A1-related glucose and galactose malabsorption	SLC5A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	SLC5A5-related hypothyroidism	SLC5A5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	SLC5A6-related neurodegenerative disorder	SLC5A6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	SLC5A7-related myasthenia	SLC5A7	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Hyperekplexia	SLC6A5-related hyperekplexia	SLC6A5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of creatine metabolism	SLC6A8-related cerebral creatine deficiency	SLC6A8	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis	SLC7A7-related lysinuric protein intolerance	SLC7A7	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Congenital diarrheal disorders	SLC9A3-related secretory diarrhea	SLC9A3	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	SMARCB1-related rhabdoid tumor predisposition syndrome and schwannomatosis spectrum	SMARCB1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	SMARCD2-related specific granule deficiency	SMARCD2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Progressive muscle weakness	SMN1-related spinal muscular atrophy	SMN1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	SNX10-related osteopetrosis	SNX10	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	SOS1-related RASopathy	SOS1	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	SOS2-related RASopathy	SOS2	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	SOX2-related multiple anomalies and neurodevelopmental spectrum disorder	SOX2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	SOX9-related campomelic dysplasia	SOX9	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	SP110-related hepatic venoocclusive disease with immunodeficiency	SP110	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	SPARC-related osteogenesis imperfecta	SPARC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	SPI1-related agammaglobulinemia	SPI1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	SPINK5-related Netherton syndrome	SPINK5	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Movement disorders	SPR-related sepiapterin reductase deficiency	SPR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	SRP54-related neutropenia	SRP54	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	SRY-related variations of sex characteristics	SRY	XL	Unknown	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Gonadal dysgenesis disorders	SRY-related variations of sex characteristics	SRY	YL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	STAR-related lipoid adrenal hyperplasia	STAR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Mendelian susceptibility to mycobacterial disease (MSMD)	STAT1-related immunodeficiency	STAT1	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to mucocutaneous candidiasis	STAT1-related immunodeficiency	STAT1	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to severe viral infection	STAT1-related immunodeficiency	STAT1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to severe viral infection	STAT2-related immunodeficiency	STAT2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Regulatory T cell defects	STAT3-related autoimmune disease	STAT3	AD	Increased function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	STAT3-related Hyper-IgE syndrome	STAT3	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other defects	STAT5B-related growth and immune dysregulation	STAT5B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	STAT6-related allergic disorder	STAT6	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Calcium channel defects	STIM1-related immunodeficiency	STIM1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	STING1-related vasculopathy	STING1	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	STING1-related vasculopathy	STING1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	STK4-related immune dysregulation	STK4	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis	STX11-related hemophagocytic lymphohistiocytosis	STX11	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	STXBP1-related neurodevelopmental disorder	STXBP1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis	STXBP2-related hemophagocytic lymphohistiocytosis	STXBP2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	SUFU-related nevoid basal cell carcinoma syndrome	SUFU	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	SYT2-related myasthenia	SYT2	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	TAFAZZIN-related Barth syndrome	TAFAZZIN	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	TAP1-related major histocompatibility complex class I deficiency	TAP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	TAP2-related major histocompatibility complex class I deficiency	TAP2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of phenylalanine and tyrosine metabolism	TAT-related tyrosinemia	TAT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	TBC1D32-related ciliopathy	TBC1D32	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Parathyroid disorders	TBCE-related neurodevelopmental disorder with multiple anomalies	TBCE	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	TBK1-related autoinflammatory disorder	TBK1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	TBL1X-related hypothyroidism	TBL1X	XL	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Thymic defects with additional congenital anomalies	TBX1-related DiGeorge syndrome	TBX1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	TBX19-related adrenocorticotrophic hormone deficiency	TBX19	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	TCIRG1-related osteopetrosis	TCIRG1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of vitamin B12 and folate metabolism	TCN2-related transcobalamin II deficiency	TCN2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Hereditary arrhythmia	TECRL-related catecholaminergic polymorphic ventricular tachycardia	TECRL	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	TERT-related telomere biology disorder	TERT	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Hereditary anemias	TF-related attransferrinemia	TF	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	TFRC-related immunodeficiency	TFRC	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	TG-related hypothyroidism	TG	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Pituitary hormone disorders	TGIF1-related holoprosencephaly spectrum disorder	TGIF1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Movement disorders	TH-related tyrosine hydroxylase deficiency	TH	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Confirmed absent in IUIS reports	THBD-related atypical hemolytic uremic syndrome	THBD	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Hematologic bone marrow failure syndromes	THPO-related amegakaryocytic thrombocytopenia	THPO	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	THRA-related hypothyroidism	THRA	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	TINF2-related telomere biology disorder	TINF2	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial nucleotide pool maintenance	TK2-related mitochondrial DNA depletion syndrome	TK2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation	TLR7-related autoimmune disorder	TLR7	XL	Increased function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of multiple glycosylation pathways	TMEM165-related congenital disorder of glycosylation (CDG IIk)	TMEM165	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	TMEM38B-related osteogenesis imperfecta	TMEM38B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of complex V subunits and assembly factors	TMEM70-related mitochondrial complex V deficiency	TMEM70	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	TNFAIP3-related autoinflammatory syndrome	TNFAIP3	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	TNFRSF11A-related osteopetrosis	TNFRSF11A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	TNFRSF11B-related hereditary hyperphosphatasia	TNFRSF11B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Non-inflammasome related conditions	TNFRSF1A-related periodic fever syndrome	TNFRSF1A	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other inborn errors of immunity related to non-hematopoietic tissues	TNFSF11-related osteopetrosis	TNFSF11	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in all serum Ig isotypes with profoundly decreased or absent B cells, agammaglobulinemia	TOP2B-related immunodeficiency and multiple anomalies	TOP2B	AD	Dominant negative	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	TP53-related Li-Fraumeni syndrome	TP53	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of thiamine metabolism	TPK1-related thiamine pyrophosphokinase deficiency	TPK1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	TPO-related hypothyroidism	TPO	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Lysosomal neurodegenerative disorder	TPP1-related ceroid lipofuscinosis	TPP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Autoimmunity with or without lymphoproliferation	TPP2-related immune dysregulation disorder	TPP2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	TRHR-related hypothyroidism	TRHR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Cardiac disorders	Congenital heart disease	Down Syndrome	Trisomy 21		Aneuploidy	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

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Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of mitochondrial transcript processing and modification	TRMU-related infantile liver failure	TRMU	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in at least 2 serum Ig isotypes with normal or low B cell numbers, CVID phenotype	TRNT1-related mitochondrial disorder	TRNT1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of manganese metabolism	TRPM6-related hypomagnesemia	TRPM6	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	TSC1-related tuberous sclerosis complex	TSC1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Epilepsies	TSC2-related tuberous sclerosis complex	TSC2	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	TSHB-related hypothyroidism	TSHB	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Thyroid disorders	TSHR-related hyperthyroidism	TSHR	AD	New function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Movement disorders	TTPA-related ataxia with vitamin E deficiency	TTPA	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Mendelian susceptibility to mycobacterial disease (MSMD)	TYK2-related immunodeficiency	TYK2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Gastrointestinal disorders	Enzymatic defect of the liver	UGT1A1-related hereditary unconjugated hyperbilirubinemia	UGT1A1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of pyrimidine metabolism	UMPS-related orotic aciduria	UMPS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Familial hemophagocytic lymphohistiocytosis	UNC13D-related hemophagocytic lymphohistiocytosis	UNC13D	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Severe reduction in serum IgG and IgA with normal/elevated IgM and normal numbers of B cells, hyper IgM	UNG-related immunodeficiency	UNG	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inherited metabolic disorders	Disorders of complex III subunits and assembly factors	UQCRC2-related mitochondrial complex III deficiency	UQCRC2	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Erythropoietic porphyrias	UROD-related hepatoerythropoietic porphyria	UROD	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

Derived from the [Bridges-NBS Condition List](#), Excluding G6PD Deficiency

Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Hematologic disorders	Erythropoietic porphyrias	UROS-related erythropoietic porphyria	UROS	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	USB1-related poikiloderma with neutropenia	USB1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	USH1C-related Usher spectrum disorder	USH1C	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	USH1G-related Usher syndrome	USH1G	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	USH2A-related Usher syndrome	USH2A	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Type 1 interferonopathies	USP18-related brain malformations spectrum disorder	USP18	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Neurologic disorders	Congenital myasthenic syndromes	VAMP1-related myasthenia	VAMP1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Rickets - vitamin D and phosphate disorders	VDR-related vitamin D-resistant rickets	VDR	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	VHL-related von Hippel-Lindau syndrome	VHL	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	VPS13B-related Cohen syndrome	VPS13B	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	VPS45-related neutropenia	VPS45	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Congenital neutropenias	WAS-related neutropenia	WAS	XL	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immunodeficiency with congenital thrombocytopenia	WAS-related Wiskott-Aldrich syndrome	WAS	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Defects of motility	WDR1-related immunodeficiency	WDR1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Renal disorders	Renal tubular acidosis	WDR72-related amelogenesis imperfecta	WDR72	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet

Sunshine Genetics Tier 1 Gene - Condition List

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Clinical Area	Disease Area	Condition	Gene	Inheritance	Mechanism	Clinician ACT Sheet	Family ACT Sheet
Hearing loss disorders	Congenital hearing loss	WHRN-related Usher syndrome	WHRN	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Immunodeficiency with congenital thrombocytopenia	WIPF1-related Wiskott-Aldrich syndrome	WIPF1	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Adrenal disorders	WNK1-related pseudohypoaldosteronism	WNK1	AD	Unknown	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Bone disorders	WNT1-related osteogenesis imperfecta	WNT1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Bone marrow failure syndromes	WRAP53-related telomere biology disorder	WRAP53	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Hereditary cancer predisposition syndromes	Childhood solid tumor predisposition syndromes	WT1-related Wilms tumor	WT1	AD	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Susceptibility to EBV and lymphoproliferative conditions	XIAP-related immune dysregulation disorder	XIAP	XL	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Dermatologic disorders	Xeroderma pigmentosum	XPC-related xeroderma pigmentosum	XPC	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Combined immunodeficiency (CID), generally less profound than SCID	ZAP70-related immunodeficiency	ZAP70	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Other DNA repair defects	ZBTB24-related immunodeficiency and multiple anomalies spectrum disorder	ZBTB24	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Endocrine disorders	Congenital hyperinsulinism, neonatal diabetes mellitus, and pancreatic beta cell disorders	ZFP57-related transient neonatal diabetes mellitus	ZFP57	AR	Unknown	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Syndromes with elevated IgE (HIES)	ZNF341-related immunodeficiency	ZNF341	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet
Inborn errors of immunity	Predisposition to severe viral infection	ZNFX1-related immunodeficiency with hematological anomalies	ZNFX1	AR	Loss of function	Clinician ACT Sheet	Family ACT Sheet